

WHAT IS CLAIMED IS:

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1 11. The isolated nucleic acid molecule of claim 5,
2 wherein the sequence of the nucleic acid molecule is identical
3 to that of Figure 2.

1 12. A polypeptide encoded by the nucleic acid
2 molecule of claim 5.

1 13. An antibody which specifically recognizes the
2 polypeptide of claim 12.

1 14. A method to determine the presence or absence
2 of the common hereditary hemochromatosis (HH) gene mutation in
3 an individual comprising:

4 providing DNA or RNA from the individual; and
5 assessing the DNA or RNA for the presence or
6 absence of a haplotype of Table 1,

7 wherein, as a result, the absence of a haplotype of
8 Table 1 indicates the likely absence of the HH gene mutation
9 in the genome of the individual and the presence of the
10 haplotype indicates the likely presence of the HH gene
11 mutation in the genome of the individual.

1 15. The method of claim 14, wherein the method
2 further comprises assessing the RNA or DNA for the presence of
3 24d1 and/or 24d2.

1 16. The method of claim 14, wherein the method
2 further comprises assessing the RNA or DNA for the presence of
3 at least one of polymorphisms HHP-1, HHP-19, or HHP-29, or
4 microsatellite repeat alleles 19D9:205; 18B4:235; 1A2:239;
5 1E4:271; 24E2:245; 2B8:206; 3321-1:98; 4073-1:182; 4440-1:180;
6 4440-2:139; 731-1:177; 5091-1:148; 3216-1:221; 4072-2:170;
7 950-1:142; 950-2:164; 950-3:165; 950-4:128; 950-6:151; 950-
8 8:137; 63-1:151; 63-2:113; 63-3:169; 65-1:206; 65-2:159; 68-
9 1:167; 241-5:108; 241-29:113; 373-8:151; and 373-29:113,
10 D6S258:199, D6S265:122, D6S105:124; D6S306:238; D6S464:206; or
11 D6S1001:180.

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1 17. The method of claim 14, wherein the haplotype
2 comprises at least two polymorphic sites of Table 1.

1 18. The method of claim 17, wherein one of the at
2 least two polymorphic sites of Table 1 is at base 35983 or
3 61465 of Figure 1.

1 19. The method of claim 13, wherein the haplotype
2 comprises at least three polymorphic sites of Table 1.

IMPROPER
DEPENDENCY

1 20. A method to determine the presence or absence
2 of the common hereditary hemochromatosis (HH) gene mutation in
3 an individual comprising:
4 providing DNA or RNA from the individual; and
5 assessing the DNA or RNA for the presence or
6 absence of a genotype defined by a polymorphic allele of Table
7 1,

8 wherein, as a result, the absence of a genotype
9 defined by a polymorphic allele of Table 1 indicates the
10 likely absence of the HH gene mutation in the genome of the
11 individual and the presence of the genotype indicates the
12 likely presence of the HH gene mutation in the genome of the
13 individual.

1 21. The method of claim 20, wherein the polymorphic
2 allele occurs in less than about 50% of a random population of
3 individuals.

1 22. The method of claim 20, wherein the polymorphic
2 allele occurs in less than about 25% of a random population of
3 individuals.

1 23. The method of claim 20, wherein the polymorphic
2 allele occurs in less than about 5% of a random population of
3 individuals.

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- 1 24. The method of claim 20, wherein the genotype is
2 C182.1G7C.
- 1 25. The method of claim 20, wherein the genotype is
2 C195.1H5T.
- 1 26. A kit comprising one or more oligonucleotides
2 of claim 1.
- 1 27. A kit comprising at least one oligonucleotide
2 pair of claim 4.
- 1 28. A culture of lymphoblastoid cells having the
2 designation HC14.

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